

Figure 2: Components of Evaluation and Care Reference Card

Components of Evaluation and Care Reference Card American College of Medical Genetics	Components of Evaluation and Care Reference Card New York State Department of Health
<p>Evaluation of the Newborn with Malformations Components of Evaluation and Care History</p> <ul style="list-style-type: none"> • Prenatal <ul style="list-style-type: none"> • Maternal age, parity, health • Complications of pregnancy • Illnesses and treatments • Exposures (drug, etc) • Folic acid supplementation • Prenatal testing • Perinatal <ul style="list-style-type: none"> • Duration of pregnancy • Intrapartum course • Presentation and mode of delivery • Complications and course • Family history <ul style="list-style-type: none"> • Information re: three generations • Malformed infants • Stillborns • Familial disorders or traits • Consanguinity • Ethnic background <p>Physical Examination</p> <ul style="list-style-type: none"> • Gestational age, growth parameters and measurements (feet) • Comprehensive examination <ul style="list-style-type: none"> • General appearance <ul style="list-style-type: none"> - Proportionality and symmetry - Positioning, posture - Behavior - Other observations • Skin pigmentation, dimples, peeling, vascular or other lesions • Head shape, symmetry, fontanelles • Scalp hair parting and whorls • Facial features <ul style="list-style-type: none"> - Pupils, orbits, palpebral fissure length and slant - Ear rotation, size and shape - Nasal appearance and patency - Lips, philtrum and vermillion border - Palate, tongue and alveolar ridges - Mandibular shape and size • Nuchal posterior hairline, redundant skin or webbing, sinus tracts • Chest shape, symmetry, location of nipples, +/- accessory nipples • Heart murmurs and pulses • Lung sound symmetry 	<p>Physical Examination (continued)</p> <ul style="list-style-type: none"> • Abdominal wall and palpation abnormalities, e.g. masses • Genital appearance and maturation • Anal orifice location and patency • Back symmetry +/- sinus tracts or hair tufts in interspinous cleft • Extremity proportion, appearance, range of motion, abnormalities <ul style="list-style-type: none"> - Palmar creases, flexion creases at joints • Neurological exam, including tone, alertness & reflexes <p>Initial Impression & Differential Diagnosis</p> <ul style="list-style-type: none"> • Single (isolated) malformation • Multiple malformations of known pattern (syndrome identified) • Multiple malformations (pattern not recognized) <p>Diagnostic Evaluation</p> <ul style="list-style-type: none"> • Imaging • Chromosomal analysis • Other genetic tests • Consider specialty referral and/or consultation <p>Develop Working Diagnosis and Counsel Family</p> <p>Counseling the Family</p> <ul style="list-style-type: none"> • Counseling principles <ul style="list-style-type: none"> • Supportive • Respect privacy and confidentiality • Facilitate grieving process • Supportive setting <ul style="list-style-type: none"> • Privacy and quiet • Inclusion of others according to family preference • Content <ul style="list-style-type: none"> • Medical facts and diagnoses • Etiology and prognosis • Treatment plan and priorities • Uncertainties • Resources • Recurrence risks • Family psychosocial support <p>Longitudinal Care and Case Management</p> <ul style="list-style-type: none"> • Written reports and written materials for families • Ongoing care and referrals as indicated